National Human Genome Research Institute
Advancing human health through genomics research

GENOMIC HEALTHCARE BRANCH (GHB)

Standards: HL7 FH data model and core data elements - status

GRANT M. WOOD

INTERMOUNTAIN HEALTHCARE CLINICAL GENETICS INSTITUTE

Annual Savings at Highest Level of Interoperability

Interoperability Between Hospital-Based Outpatient Clinicians and External Laboratories - $31.8 billion

Connectivity Between Office-Based Clinicians and External Radiology Centers - $26.2 billion

Interoperability Between Outpatient Providers and Pharmacies - $2.71 billion

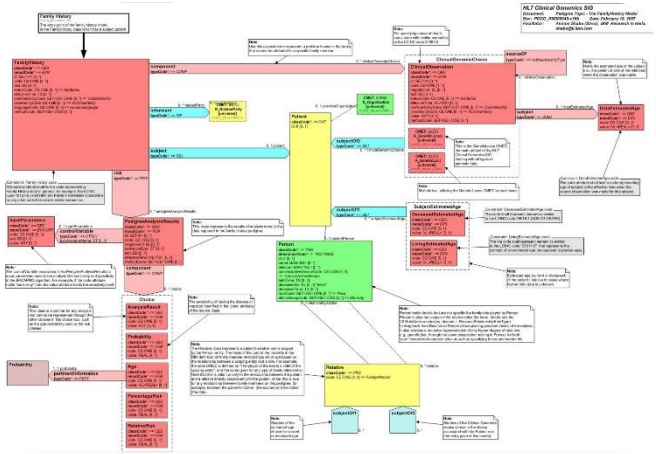
Provider to Provider Connectivity - $13.2 billion

If a medium-size hospital would invest $2.7 million, after the first year, spending $250,000 per year to maintain those systems, it would accrue benefits of $1.3 million per year

Walker, et al. The Value Of Health Care Information Exchange And Interoperability *Health Affairs* Web Exclusive, January 19, 2005

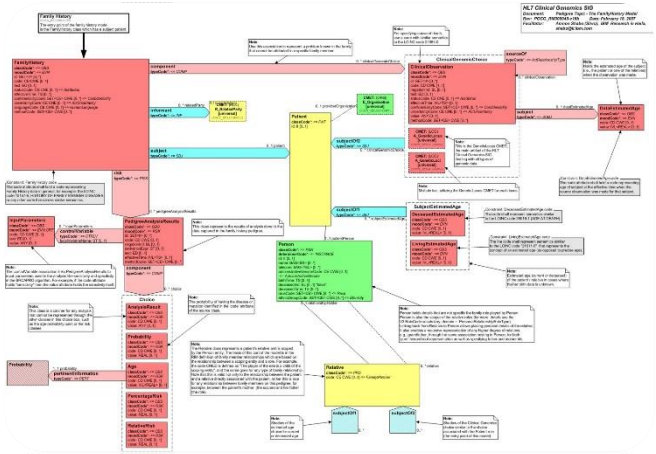
HL7 Intro – Value of Standards

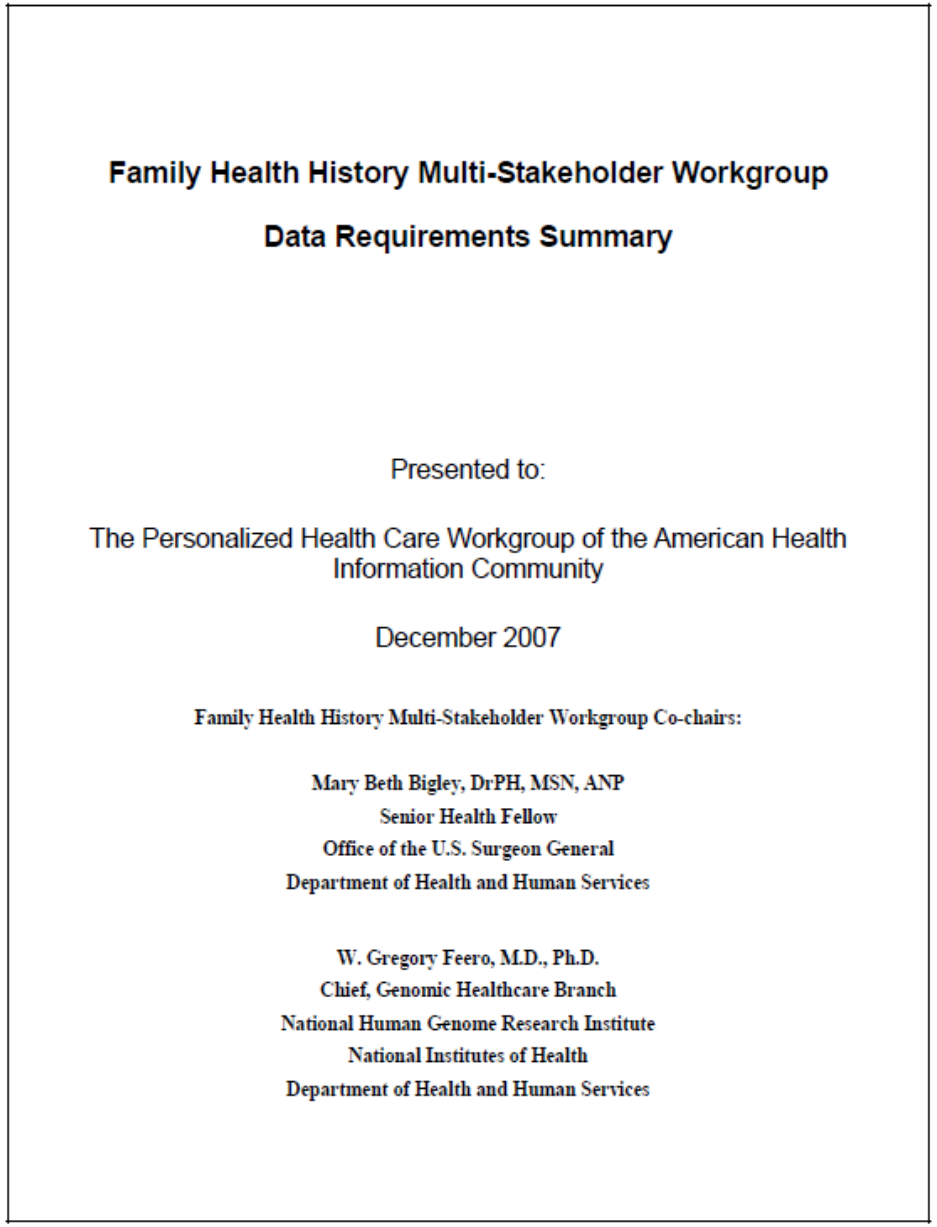
- Family health history can be used as a clinical screening tool to promote preventive medicine
- It has been demonstrated that familial risk stratification is feasible, that rational strategies exist for familial risk-tailored screening and prevention, and that such interventions lead to improved health outcomes
- Several familial risk-based guidelines have been put forth by medical societies



HL7 Intro – Value of Standards

- 2007, 2013 - Family Health History or Pedigree model
- 2009 to 2013 - Genetic Variation model and Cytogenetics model for laboratory reporting of genetic test results to the EHR
- 2011 to 2013 - Clinical Document Architecture (CDA) Genetic Test Results electronic document
- 2014 and beyond – Fast Health Information Resource (FHIR) for both family health history and genetic/genomic testing





Family Health History Multi-Stakeholder Workgroup

Data Requirements Summary

Presented to:

The Personalized Health Care Workgroup of the American Health Information Community

December 2007

Family Health History Multi-Stakeholder Workgroup Co-chairs:

Mary Beth Bigley, DrPH, MSN, ANP
Senior Health Fellow
Office of the U.S. Surgeon General
Department of Health and Human Services

W. Gregory Feero, M.D., Ph.D.
Chief, Genomic Healthcare Branch
National Human Genome Research Institute
National Institutes of Health
Department of Health and Human Services

AHIC Recommendations

A multi-stakeholder workgroup, including the private sector, federal health care providers, and federal Public Health Service agencies, should be formed to develop a core minimum data set and common data definition available for primary care collection of family health history information.

On July 31, 2007, the Personalized Health Care (PHC) Workgroup (http://www.hhs.gov/healthit/ahic/healthcare/) submitted a set of recommendations to the America Health Information Community (AHIC). These recommendations, subsequently adopted by AHIC, were aimed at enhancing the integration of interoperable family health history information into Electronic Health Records (EHRs).

HL7 Intro – Value of Standards

Hughes RiskApps™

MeTree

PROGENY

My Family Health Portrait
A tool from the Surgeon General

Intermountain

FamilyHistoryTool™

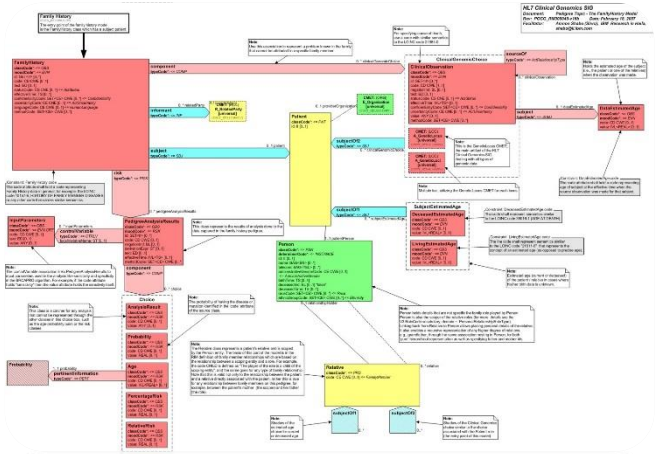
Counsyl

INVITAE

Genomic Repository

EHR

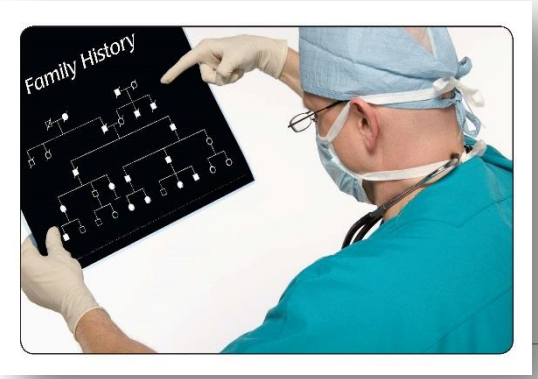
Family Health History



HL7 Intro – Value of Standards

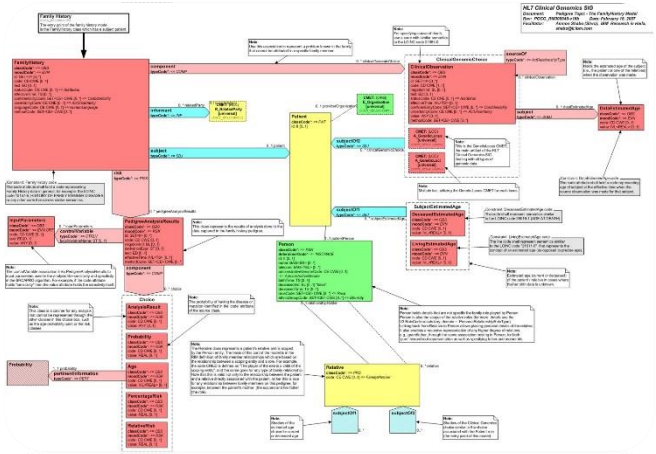
A New Paradigm for FHH

Supporting an ecosystem of tools
and their different users,
shared data repositories,
and clinical systems



HL7 V3 Review

1) Record information
2) Person of focus (Proband)
3) Mother & Father IDs, family relationship codes
4) Age of person / death date
5) Disease or condition
6) Age of disease onset / age of disease death
7) SNOMED, LOINC codes for #4, #5, and #6
8) Risk analysis
9) Sequence variant / Genetic mutation minimal core dataset

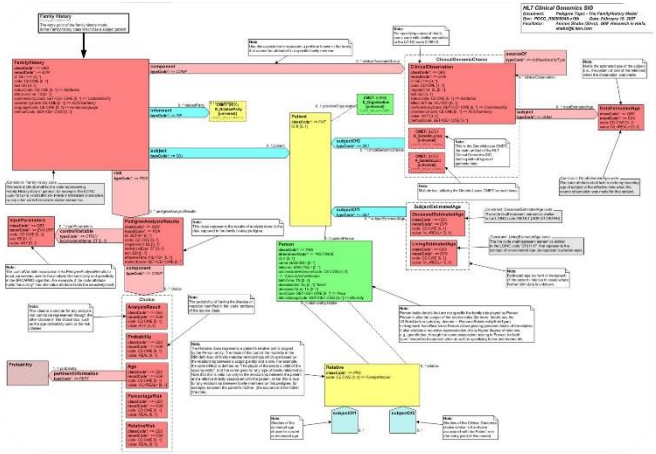


HL7 FHIR Resources and Profiles

HL7 FHIR Family Member History for Genetics Analysis

http://hl7.org/fhir/2016May/familymemberhistory.html

http://hl7.org/fhir/2016May/family-member-history-genetics.html



This is a pre-release of a future version of FHIR (expected to be STU 3). The current version is DSTU 2.
For a full list of available versions, see the Directory of published versions.

4.41.9 Family member history for genetics analysis

Adds additional information to a family member history supporting both the capture of mother/father relationships as well as additional observations necessary to enable genetics-based risk analysis for patients

4.41.9.1 Content

Profiles:

FamilyMemberHistory-Genetic	Adds additional information to a family member history supporting both the capture of mother/father relationships as well as additional observations necessary to enable genetics-based risk analysis for patients

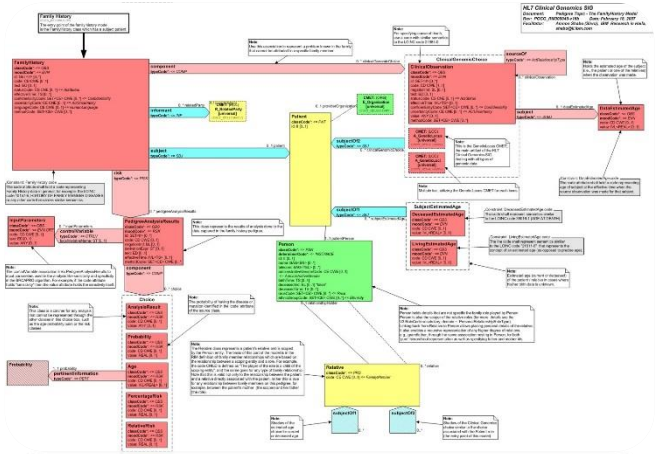
Extensions:

family-member-history-genetics-parent	**Mother(s) & Father(s) - genetic & other** : Identifies a parent of the relative.
family-member-history-genetics-observation	**Genetic markers, ethnicity, etc.** : Allows capturing risk-relevant observations about the relative that aren't themselves a specific health condition; e.g. Certain ethnic ancestries that are disease-relevant, presence of particular genetic markers, etc.

HL7 FHIR Tool Demo

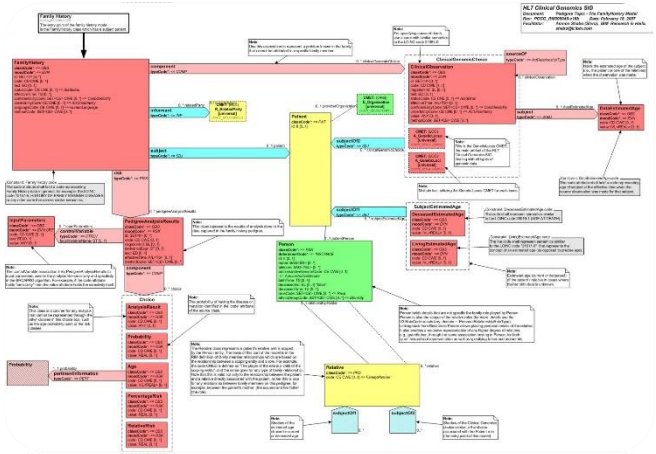
FHIR Family Member History resource

http://clinfhir.com/



FHH Consent System Concept

How would a FHH consent management system look to the patient



Assumptions – data collection standards

A family health history that includes –

- a full pedigree with coded relationships (could be HL7, SNOMED),
- coded diseases and conditions (SNOMED CT),
- coded age of onset and age of death,
- and conforms to AHIC core minimum data set

Global Alliance for Genomics & Health

Assumptions – data locations

A shared database accessible by –

- healthcare consumer and their relatives
- health providers via electronic health record
- researchers as part of a study or trial

Global Alliance for Genomics & Health

Assumptions – reasons for sharing

Healthcare consumer wants to (in full or in part) consent to sharing for the following purposes –

- To build a FHH with relatives updating the same record
- To link health records with relatives to automate the creation of a validated clinical FHH
- To have family participation in studies and trials

Global Alliance for Genomics & Health

Consent Menu Example

Conditions

- All
- Genetic only
- Show only (list)
- Hide all, but allow for use by risk scoring
- Hide all, and do not link for anything

Global Alliance for Genomics & Health

Consent Menu Example

All
- Show all conditions that would be shared

Then –
- All relatives
- All relatives except (list)
- Only these relatives (list)

Then –
- All healthcare providers, studies, trials
- All healthcare providers, studies, trials except (list)
- Only healthcare providers, studies, trials (list)

Global Alliance for Genomics & Health

Consent Menu Example

Genetic only –

- Show only what conditions could be genetic related

Then –

- All relatives
- All relatives except (list)
- Only these relatives (list)

Then –

- All healthcare providers, studies, trials
- All healthcare providers, studies, trials except (list)
- Only healthcare providers, studies, trials (list)

Global Alliance for Genomics & Health

Consent Menu Example

Show only –

- Show only conditions selected from list

Then –

- All relatives
- All relatives except (list)
- Only relatives (list)

Then –

- All healthcare providers, studies, trials
- All healthcare providers, studies, trials except (list)
- Only healthcare providers, studies, trials (list)

Global Alliance for Genomics & Health

Connecting Your Family For Better Health

Our Family Health is a new tool from Intermountain Healthcare, built to help you understand the diseases that run in your family. Get started now!

Use this new program to:

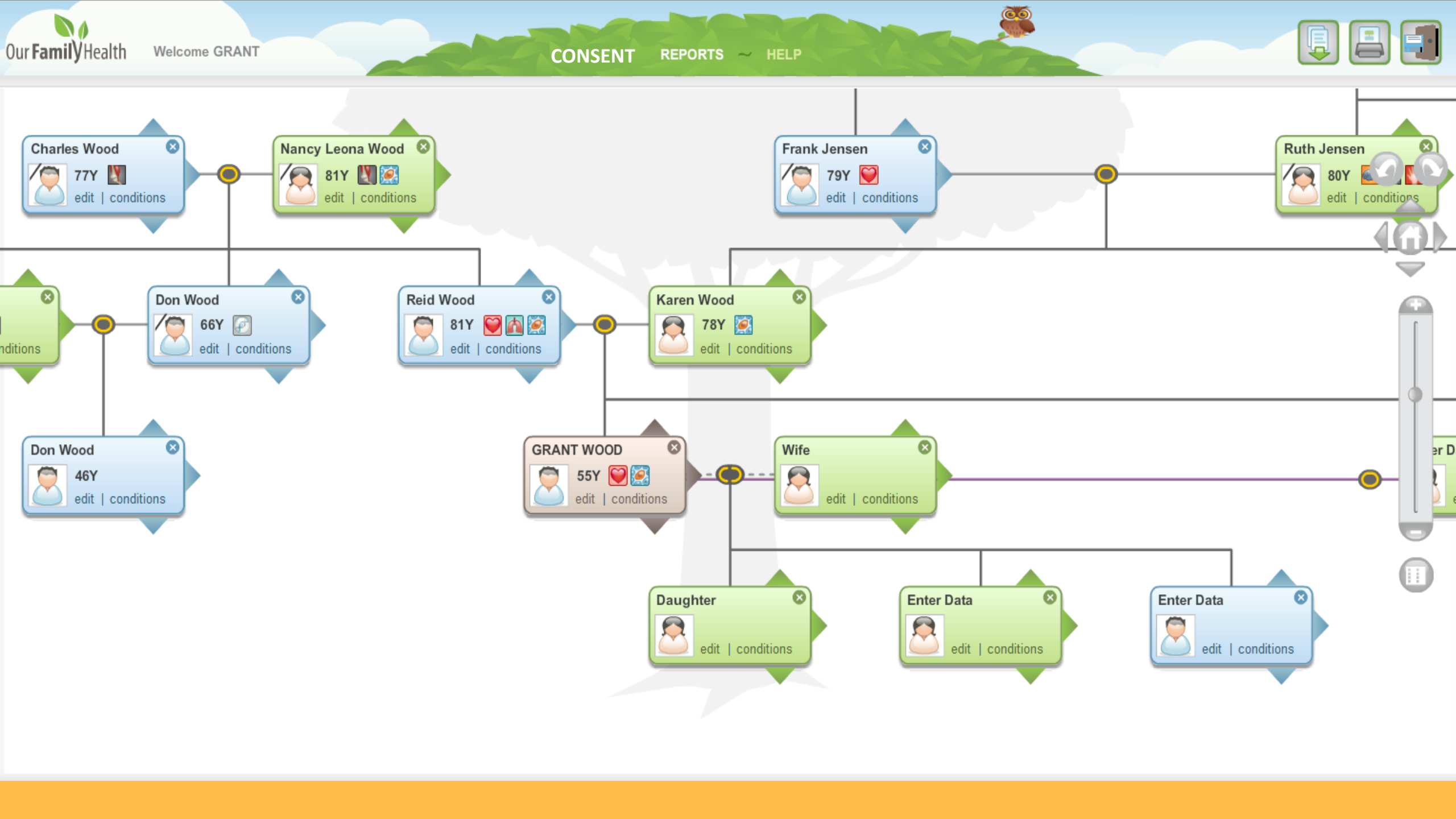
- Build your family tree.
- Add health and disease information to each person.
- Share with your family members and doctor.

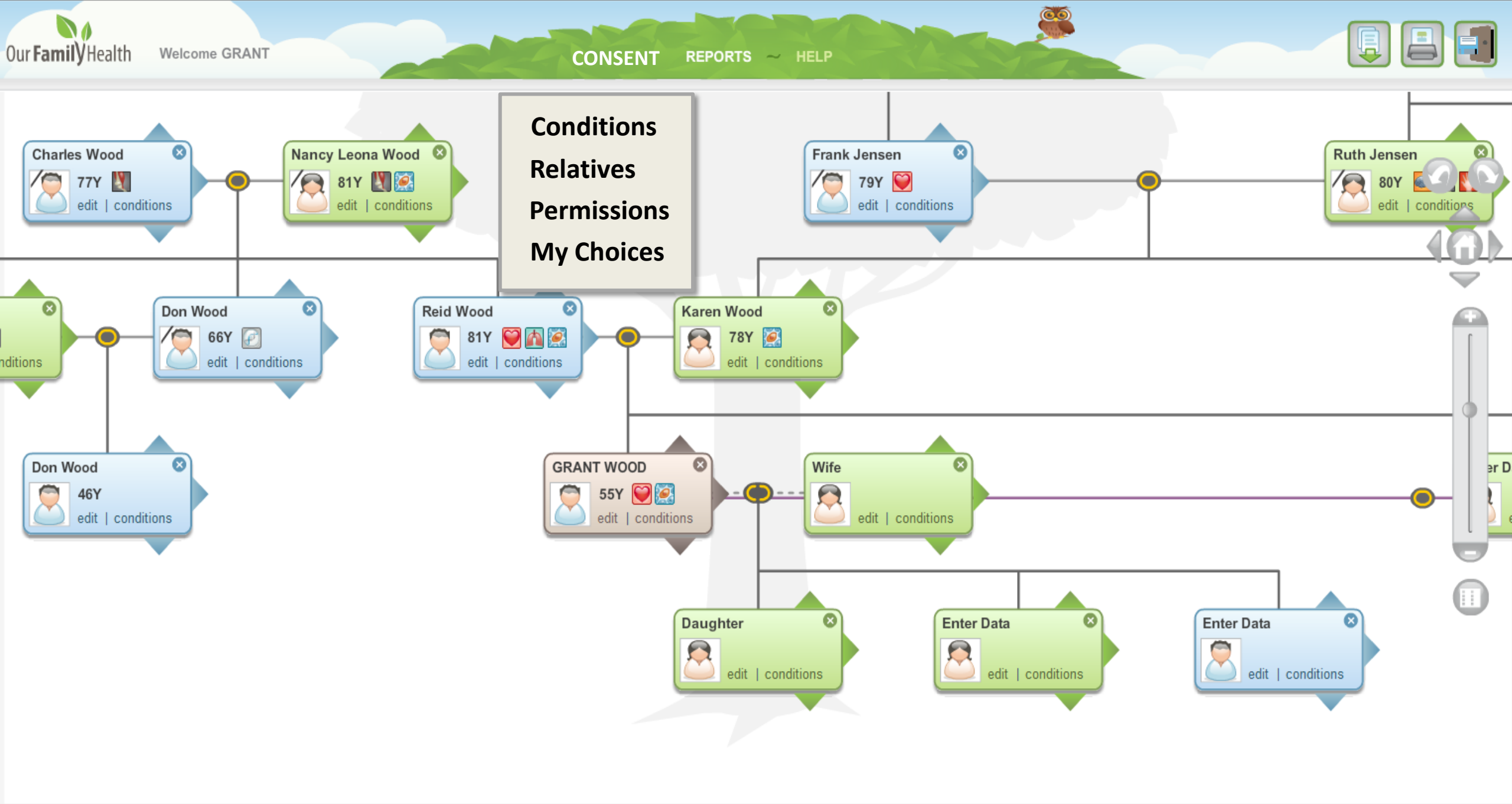
What is family health history?

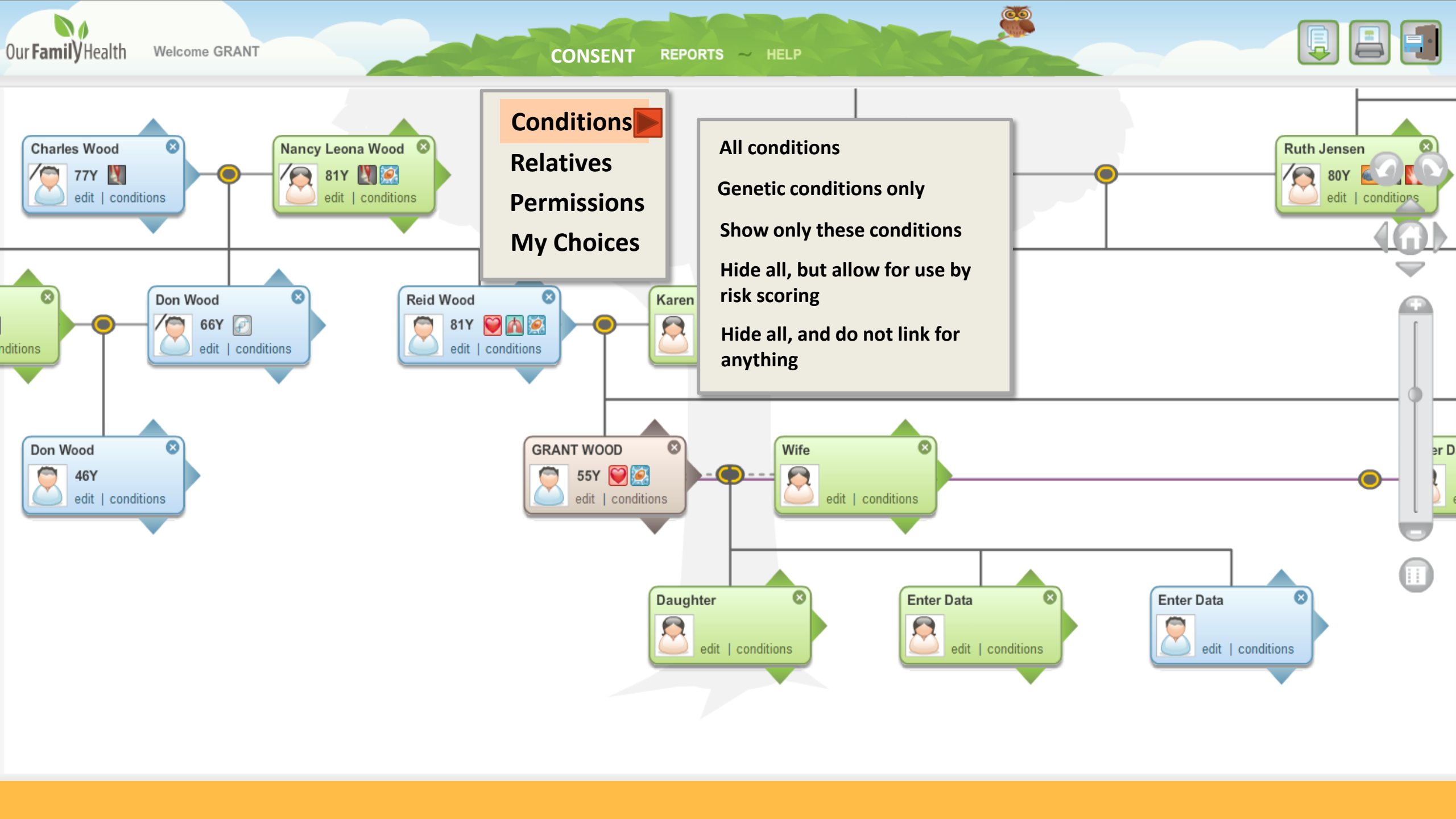
Your family health history is a collection of information about diseases that run in your family. It also includes the eating habits, activities, and environments that your family shares.

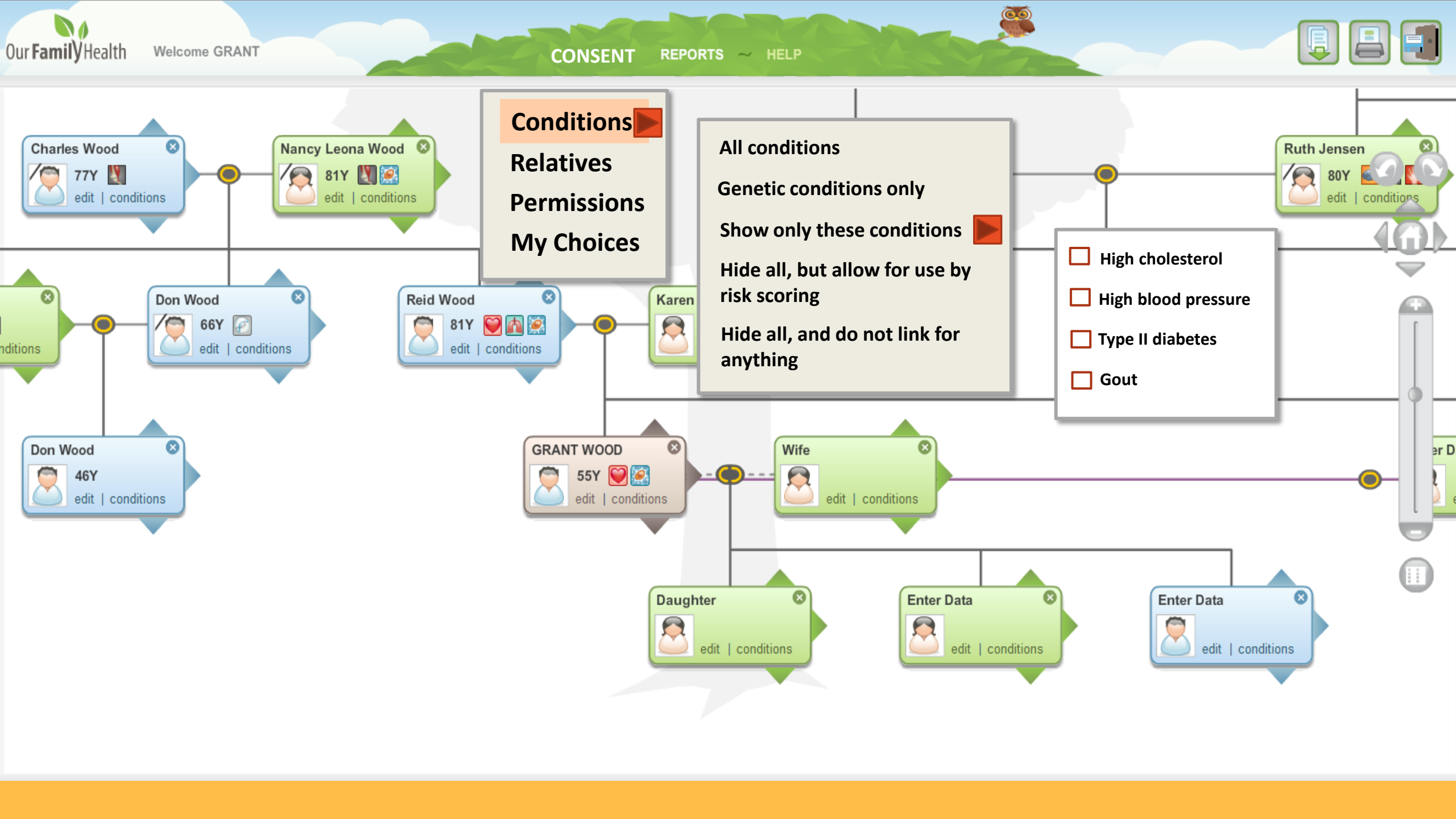
How can family health history affect my health?

Our Family Health


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
79Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen Wood
78Y
edit | conditions
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
79Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen Wood
78Y
edit | conditions
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
All conditions
Genetic conditions only
Show only these conditions
Hide all, but allow for use by risk scoring
Hide all, and do not link for anything
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
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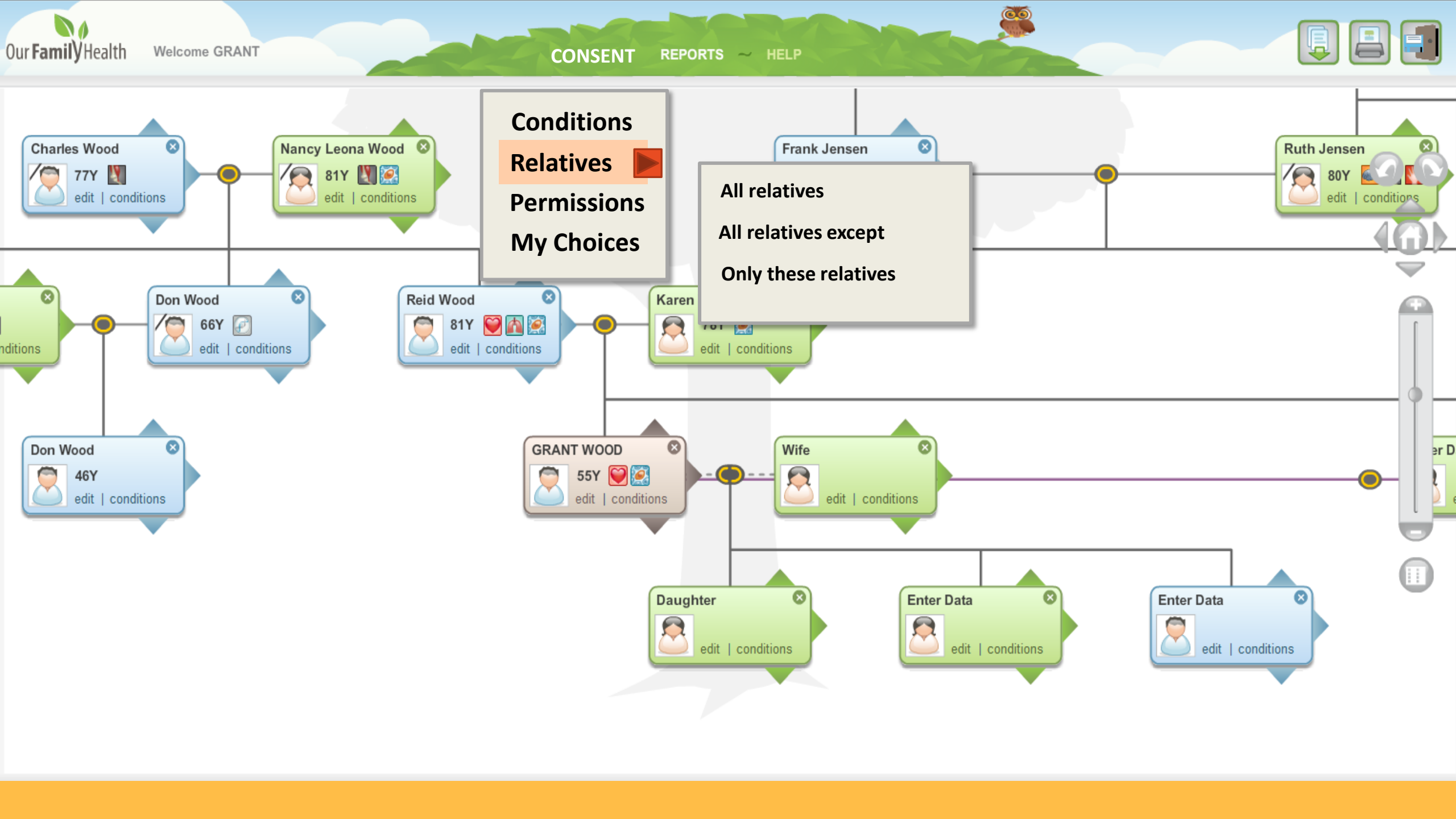
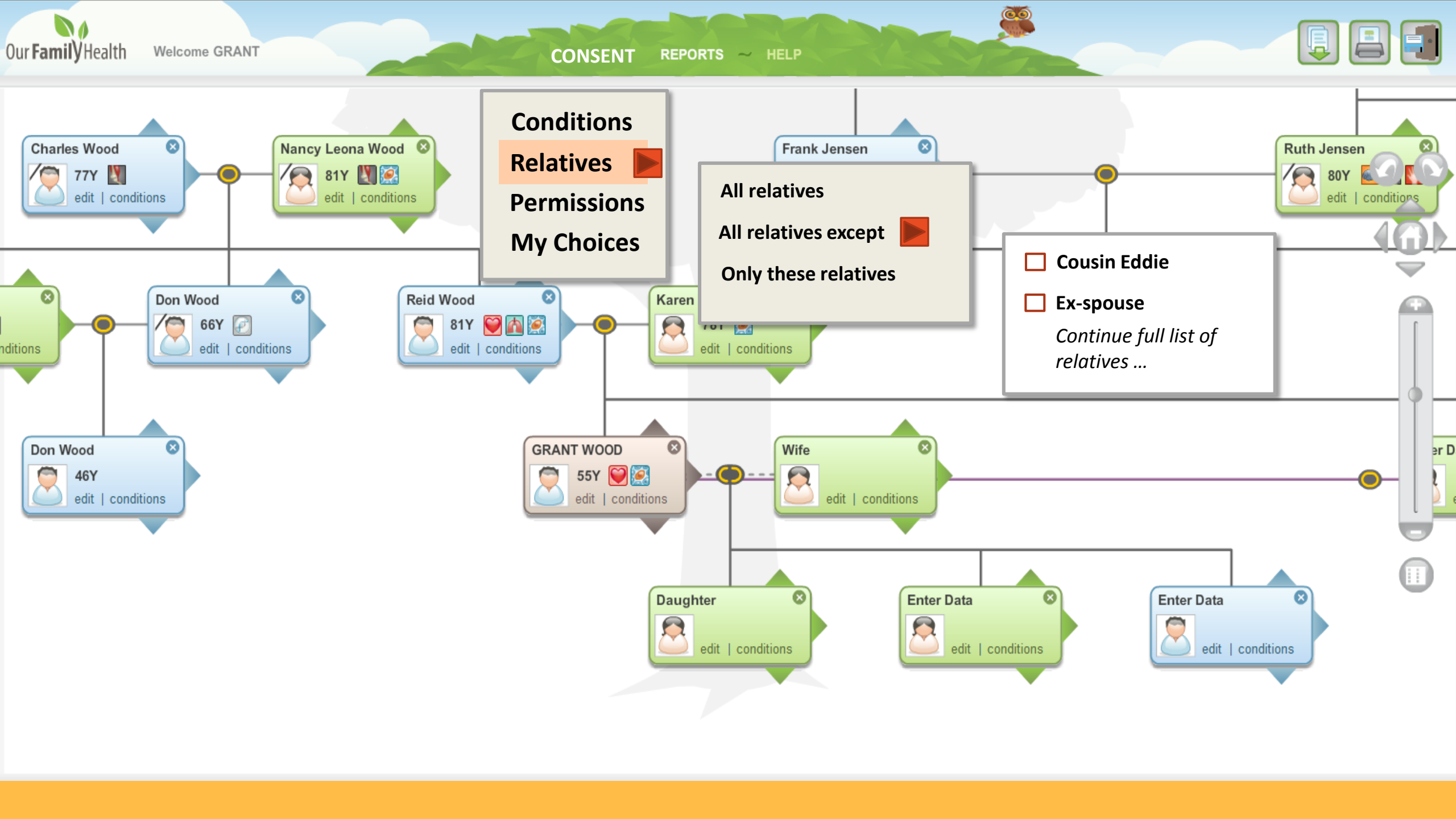
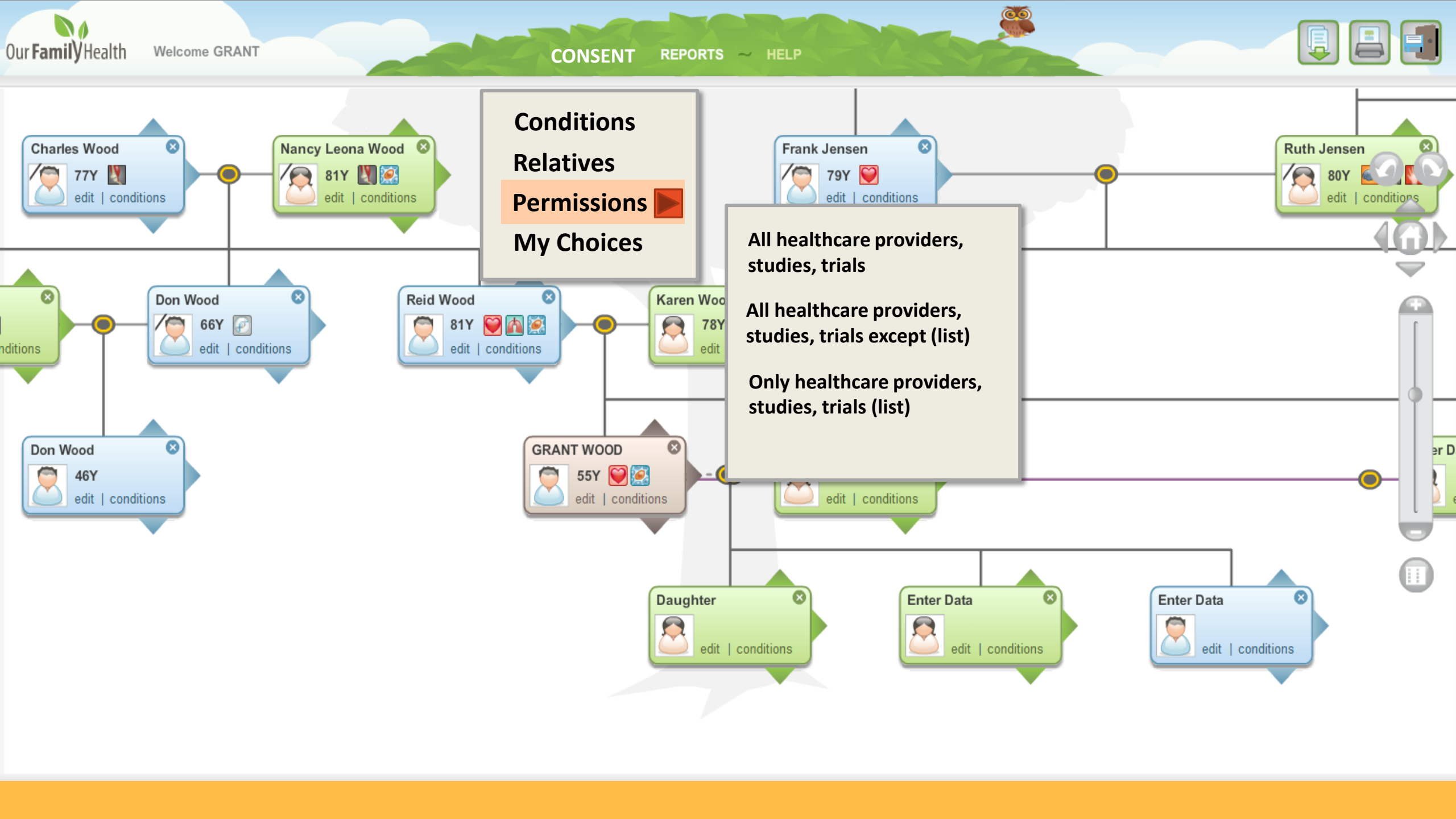
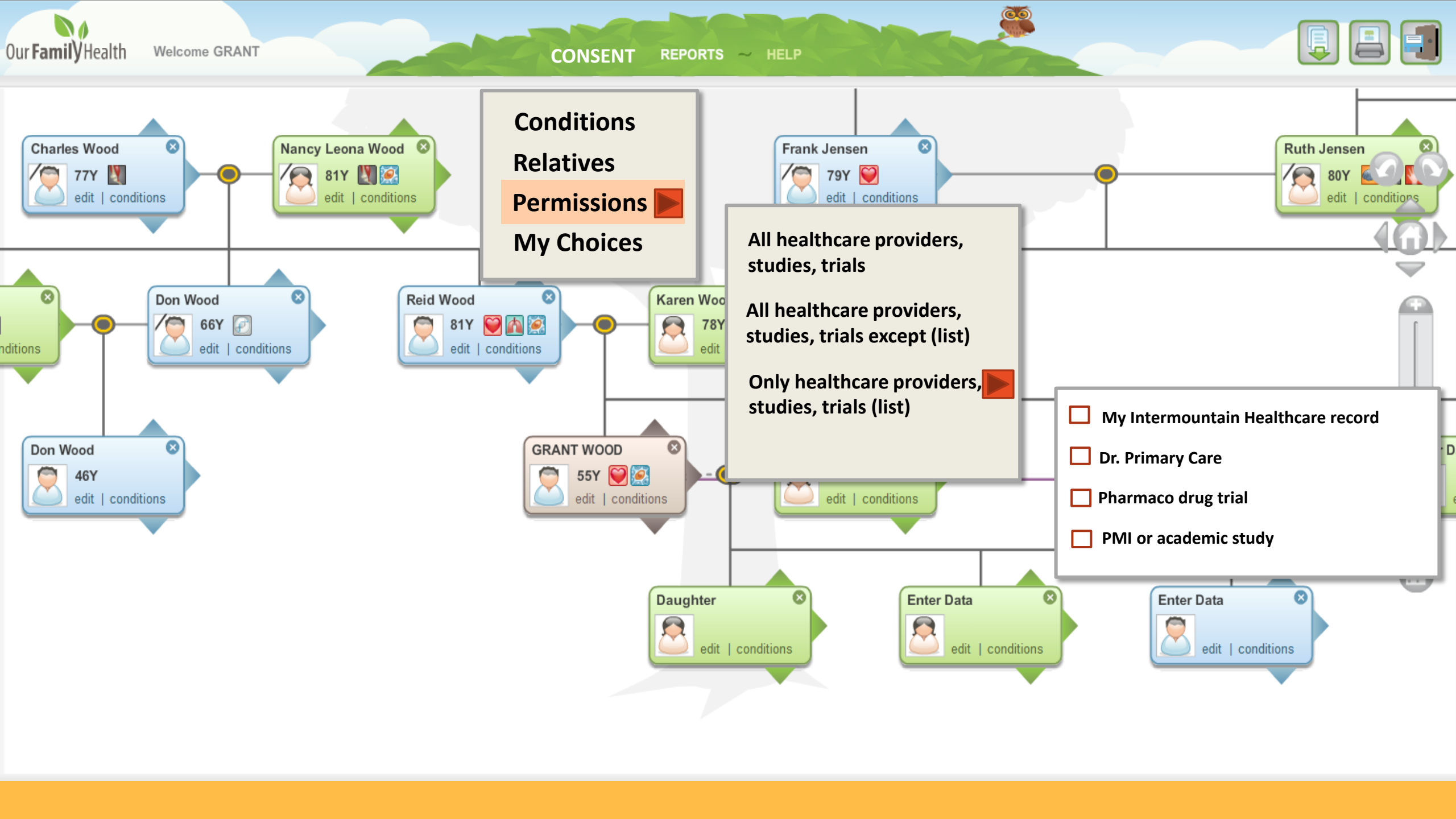
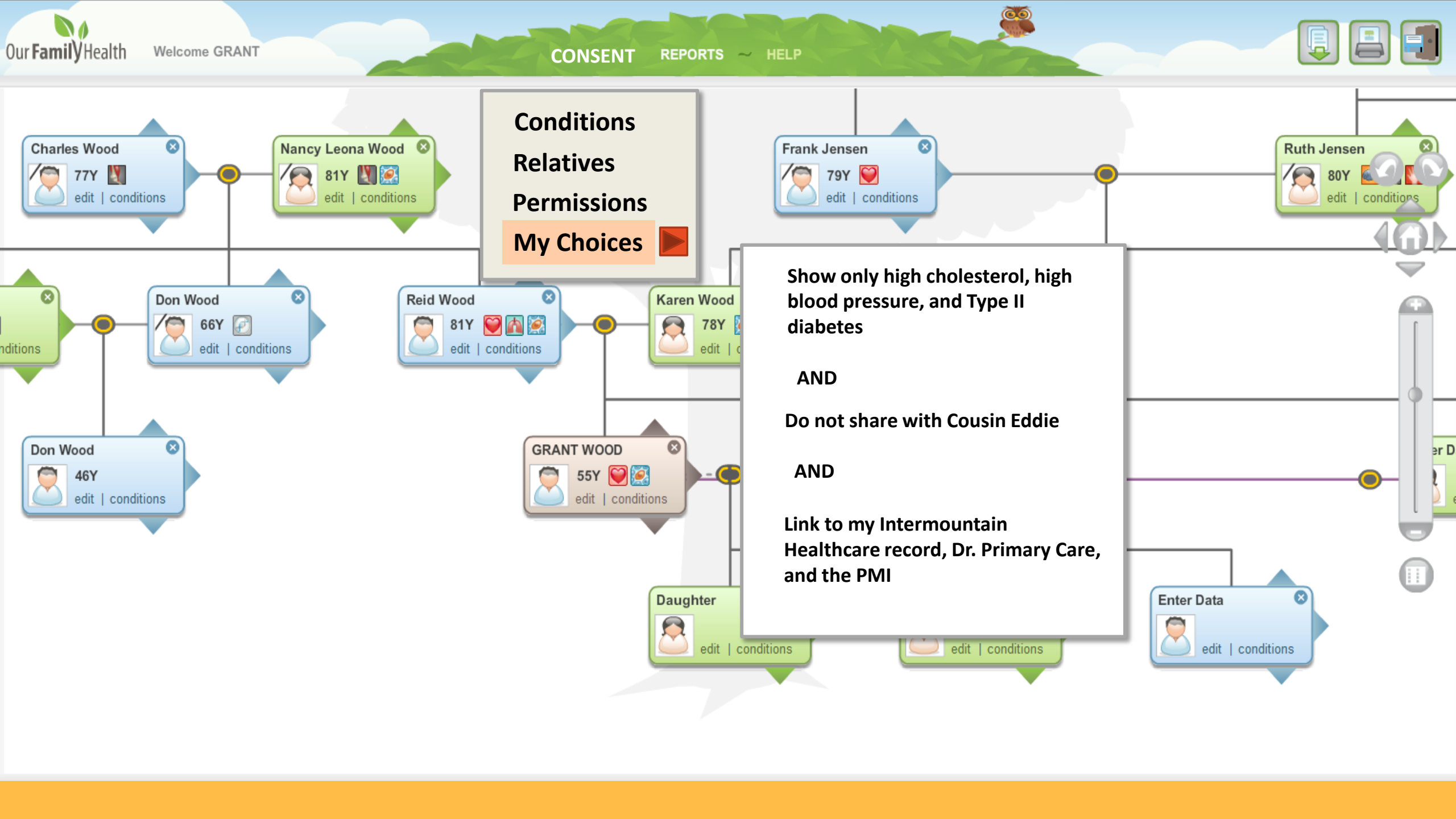
HELP
Conditions
Relatives
Permissions
My Choices
All conditions
Genetic conditions only
Show only these conditions
Hide all, but allow for use by risk scoring
Hide all, and do not link for anything
High cholesterol
High blood pressure
Type II diabetes
Gout
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our FamilyHealth
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
All relatives
All relatives except
Only these relatives
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen
edit | conditions
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our FamilyHealth
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
All relatives
All relatives except
Only these relatives
Cousin Eddie
Ex-spouse
Continue full list of relatives …
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen
edit | conditions
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Wife
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
All healthcare providers, studies, trials
All healthcare providers, studies, trials except (list)
Only healthcare providers, studies, trials (list)
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
79Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen Woo
78Y
edit
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
All healthcare providers, studies, trials
All healthcare providers, studies, trials except (list)
Only healthcare providers, studies, trials (list)
My Intermountain Healthcare record
Dr. Primary Care
Pharmaco drug trial
PMI or academic study
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
79Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen Woo
78Y
edit
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
edit | conditions
Daughter
edit | conditions
Enter Data
edit | conditions
Enter Data
edit | conditions


Our Family Health
Welcome GRANT
CONSENT
REPORTS
HELP
Conditions
Relatives
Permissions
My Choices
Show only high cholesterol, high blood pressure, and Type II diabetes
AND
Do not share with Cousin Eddie
AND
Link to my Intermountain Healthcare record, Dr. Primary Care, and the PMI
Charles Wood
77Y
edit | conditions
Nancy Leona Wood
81Y
edit | conditions
Frank Jensen
79Y
edit | conditions
Ruth Jensen
80Y
edit | conditions
Don Wood
66Y
edit | conditions
Reid Wood
81Y
edit | conditions
Karen Wood
78Y
edit
Don Wood
46Y
edit | conditions
GRANT WOOD
55Y
edit | conditions
Daughter
edit | conditions
edit | conditions
Enter Data
edit | conditions

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Hughes RiskApps™

MeTree

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A tool from the Surgeon General

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